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List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

83 papers	27,244 citations	42 h-index	95 g-index
95 ext. papers	32,491 ext. citations	19.9 avg, IF	7.25 L-index

#	Paper	IF	Citations
83	In Search of Complex Disease Risk through Genome Wide Association Studies. <i>Mathematics</i> , 2021 , 9, 3083	2.3	
82	GA4GH: International policies and standards for data sharing across genomic research and healthcare.. <i>Cell Genomics</i> , 2021 , 1, 100029-100029		20
81	Recessive Genome-wide Meta-analysis Illuminates Genetic Architecture of Type 2 Diabetes. <i>Diabetes</i> , 2021 ,	0.9	0
80	TIGER: The gene expression regulatory variation landscape of human pancreatic islets. <i>Cell Reports</i> , 2021 , 37, 109807	10.6	5
79	The impact of non-additive genetic associations on age-related complex diseases. <i>Nature Communications</i> , 2021 , 12, 2436	17.4	10
78	Polymorphic Inversions Underlie the Shared Genetic Susceptibility of Obesity-Related Diseases. <i>American Journal of Human Genetics</i> , 2020 , 106, 846-858	11	3
77	Genomic and epigenomic insights into the origin, pathogenesis, and clinical behavior of mantle cell lymphoma subtypes. <i>Blood</i> , 2020 , 136, 1419-1432	2.2	53
76	Pan-cancer analysis of whole genomes identifies driver rearrangements promoted by LINE-1 retrotransposition. <i>Nature Genetics</i> , 2020 , 52, 306-319	36.3	122
75	Extrachromosomal circular DNA drives oncogenic genome remodeling in neuroblastoma. <i>Nature Genetics</i> , 2020 , 52, 29-34	36.3	70
74	Enhancer hijacking determines extrachromosomal circular MYCN amplicon architecture in neuroblastoma. <i>Nature Communications</i> , 2020 , 11, 5823	17.4	34
73	Retrospective evaluation of whole exome and genome mutation calls in 746 cancer samples. <i>Nature Communications</i> , 2020 , 11, 4748	17.4	10
72	Maternal and fetal genetic effects on birth weight and their relevance to cardio-metabolic risk factors. <i>Nature Genetics</i> , 2019 , 51, 804-814	36.3	181
71	Insight into genetic predisposition to chronic lymphocytic leukemia from integrative epigenomics. <i>Nature Communications</i> , 2019 , 10, 3615	17.4	19
70	Human pancreatic islet three-dimensional chromatin architecture provides insights into the genetics of type 2 diabetes. <i>Nature Genetics</i> , 2019 , 51, 1137-1148	36.3	111
69	and hijack immunoglobulin light-chain enhancers in cyclin D1 mantle cell lymphoma. <i>Blood</i> , 2019 , 133, 940-951	2.2	48
68	Genomic profiling in advanced stage non-small-cell lung cancer patients with platinum-based chemotherapy identifies germline variants with prognostic value in SMYD2. <i>Cancer Treatment and Research Communications</i> , 2018 , 15, 21-31	2	8
67	Re-analysis of public genetic data reveals a rare X-chromosomal variant associated with type 2 diabetes. <i>Nature Communications</i> , 2018 , 9, 321	17.4	50

66	Genome-wide association and HLA fine-mapping studies identify risk loci and genetic pathways underlying allergic rhinitis. <i>Nature Genetics</i> , 2018 , 50, 1072-1080	36.3	52
65	Genome-wide association study meta-analysis identifies five new loci for systemic lupus erythematosus. <i>Arthritis Research and Therapy</i> , 2018 , 20, 100	5.7	47
64	A cancer-associated polymorphism in ESCRT-III disrupts the abscission checkpoint and promotes genome instability. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018 , 115, E8900-E8908	11.5	28
63	Multitrait genome association analysis identifies new susceptibility genes for human anthropometric variation in the GCAT cohort. <i>Journal of Medical Genetics</i> , 2018 , 55, 765-778	5.8	12
62	The reference epigenome and regulatory chromatin landscape of chronic lymphocytic leukemia. <i>Nature Medicine</i> , 2018 , 24, 868-880	50.5	103
61	A kidney-disease gene panel allows a comprehensive genetic diagnosis of cystic and glomerular inherited kidney diseases. <i>Kidney International</i> , 2018 , 94, 363-371	9.9	57
60	PGBD5 promotes site-specific oncogenic mutations in human tumors. <i>Nature Genetics</i> , 2017 , 49, 1005-1014	36.3	40
59	Dysregulation of Placental miRNA in Maternal Obesity Is Associated With Pre- and Postnatal Growth. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2017 , 102, 2584-2594	5.6	45
58	Metformin alters the gut microbiome of individuals with treatment-naïve type 2 diabetes, contributing to the therapeutic effects of the drug. <i>Nature Medicine</i> , 2017 , 23, 850-858	50.5	732
57	A Loss-of-Function Splice Acceptor Variant in <i>IS</i> Is Protective for Type 2 Diabetes. <i>Diabetes</i> , 2017 , 66, 2903-2914	29.14	32
56	The BLUEPRINT Data Analysis Portal. <i>Cell Systems</i> , 2016 , 3, 491-495.e5	10.6	71
55	The International Human Epigenome Consortium: A Blueprint for Scientific Collaboration and Discovery. <i>Cell</i> , 2016 , 167, 1145-1149	56.2	232
54	Tuning fresh: radiation through rewiring of central metabolism in streamlined bacteria. <i>ISME Journal</i> , 2016 , 10, 1902-14	11.9	31
53	Epigenomic analysis detects aberrant super-enhancer DNA methylation in human cancer. <i>Genome Biology</i> , 2016 , 17, 11	18.3	141
52	Genome-wide associations for birth weight and correlations with adult disease. <i>Nature</i> , 2016 , 538, 248-252	52.4	266
51	Non-coding recurrent mutations in chronic lymphocytic leukaemia. <i>Nature</i> , 2015 , 526, 519-24	50.4	565
50	Altered Circulating miRNA Expression Profile in Pregestational and Gestational Obesity. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2015 , 100, E1446-56	5.6	68
49	Surgery-Induced Weight Loss Is Associated With the Downregulation of Genes Targeted by MicroRNAs in Adipose Tissue. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2015 , 100, E1467-76	5.6	35

48	A comprehensive assessment of somatic mutation detection in cancer using whole-genome sequencing. <i>Nature Communications</i> , 2015 , 6, 10001	17.4	199
47	A genome-wide association study identifies CDHR3 as a susceptibility locus for early childhood asthma with severe exacerbations. <i>Nature Genetics</i> , 2014 , 46, 51-5	36.3	376
46	Comprehensive characterization of complex structural variations in cancer by directly comparing genome sequence reads. <i>Nature Biotechnology</i> , 2014 , 32, 1106-12	44.5	62
45	Somatic signature of brain-specific single nucleotide variations in sporadic Alzheimer's disease. <i>Journal of Alzheimer's Disease</i> , 2014 , 42, 1357-82	4.3	31
44	Genomics of ecological adaptation in cactophilic <i>Drosophila</i> . <i>Genome Biology and Evolution</i> , 2014 , 7, 349-56	5.6	38
43	Adaptation to environmental factors shapes the organization of regulatory regions in microbial communities. <i>BMC Genomics</i> , 2014 , 15, 877	4.5	13
42	Unravelling the hidden DNA structural/physical code provides novel insights on promoter location. <i>Nucleic Acids Research</i> , 2013 , 41, 7220-30	20.1	11
41	Common genetic variants of surfactant protein-D (SP-D) are associated with type 2 diabetes. <i>PLoS ONE</i> , 2013 , 8, e60468	3.7	12
40	Impact of methylation on the physical properties of DNA. <i>Biophysical Journal</i> , 2012 , 102, 2140-8	2.9	89
39	The tomato genome sequence provides insights into fleshy fruit evolution. <i>Nature</i> , 2012 , 485, 635-41	50.4	2138
38	BLUEPRINT to decode the epigenetic signature written in blood. <i>Nature Biotechnology</i> , 2012 , 30, 224-6	44.5	261
37	Identification of novel type 2 diabetes candidate genes involved in the crosstalk between the mitochondrial and the insulin signaling systems. <i>PLoS Genetics</i> , 2012 , 8, e1003046	6	17
36	ReLA, a local alignment search tool for the identification of distal and proximal gene regulatory regions and their conserved transcription factor binding sites. <i>Bioinformatics</i> , 2012 , 28, 763-70	7.2	11
35	Enterotypes of the human gut microbiome. <i>Nature</i> , 2011 , 473, 174-80	50.4	4240
34	Whole-genome sequencing identifies recurrent mutations in chronic lymphocytic leukaemia. <i>Nature</i> , 2011 , 475, 101-5	50.4	1206
33	Selective maintenance of <i>Drosophila</i> tandemly arranged duplicated genes during evolution. <i>Genome Biology</i> , 2008 , 9, R176	18.3	10
32	Projection structure of a member of the amino acid/polyamine/organocation transporter superfamily. <i>Journal of Biological Chemistry</i> , 2008 , 283, 33240-8	5.4	34
31	DNAlive: a tool for the physical analysis of DNA at the genomic scale. <i>Bioinformatics</i> , 2008 , 24, 1731-2	7.2	22

30	Is there selection for the pace of successive inactivation of the arpAT gene in primates?. <i>Journal of Molecular Evolution</i> , 2008 , 67, 23-8	3.1	2
29	Functional and structural characterization of the first prokaryotic member of the L-amino acid transporter (LAT) family: a model for APC transporters. <i>Journal of Biological Chemistry</i> , 2007 , 282, 13270-81	5.4	35
28	Determining promoter location based on DNA structure first-principles calculations. <i>Genome Biology</i> , 2007 , 8, R263	18.3	98
27	Identification and analysis of genes and pseudogenes within duplicated regions in the human and mouse genomes. <i>PLoS Computational Biology</i> , 2006 , 2, e76	5	20
26	PAL2NAL: robust conversion of protein sequence alignments into the corresponding codon alignments. <i>Nucleic Acids Research</i> , 2006 , 34, W609-12	20.1	1686
25	Protein coding potential of retroviruses and other transposable elements in vertebrate genomes. <i>Nucleic Acids Research</i> , 2005 , 33, 946-54	20.1	40
24	Generation and annotation of the DNA sequences of human chromosomes 2 and 4. <i>Nature</i> , 2005 , 434, 724-31	50.4	61
23	Initial sequence of the chimpanzee genome and comparison with the human genome. <i>Nature</i> , 2005 , 437, 69-87	50.4	1828
22	Identification and functional characterization of a novel low affinity aromatic-preferring amino acid transporter (arpAT). One of the few proteins silenced during primate evolution. <i>Journal of Biological Chemistry</i> , 2005 , 280, 19364-72	5.4	21
21	BLAST2GENE: a comprehensive conversion of BLAST output into independent genes and gene fragments. <i>Bioinformatics</i> , 2004 , 20, 1968-70	7.2	17
20	Genome sequence of the Brown Norway rat yields insights into mammalian evolution. <i>Nature</i> , 2004 , 428, 493-521	50.4	1689
19	Sequence and comparative analysis of the chicken genome provide unique perspectives on vertebrate evolution. <i>Nature</i> , 2004 , 432, 695-716	50.4	2143
18	A genome-wide survey of human pseudogenes. <i>Genome Research</i> , 2003 , 13, 2559-67	9.7	213
17	The DNA sequence of human chromosome 7. <i>Nature</i> , 2003 , 424, 157-64	50.4	202
16	Basolateral LAT-2 has a major role in the transepithelial flux of L-cystine in the renal proximal tubule cell line OK. <i>Journal of the American Society of Nephrology: JASN</i> , 2003 , 14, 837-47	12.7	39
15	The human genome: genes, pseudogenes, and variation on chromosome 7. <i>Cold Spring Harbor Symposia on Quantitative Biology</i> , 2003 , 68, 13-22	3.9	
14	Initial sequencing and comparative analysis of the mouse genome. <i>Nature</i> , 2002 , 420, 520-62	50.4	5376
13	Comparative genome and proteome analysis of <i>Anopheles gambiae</i> and <i>Drosophila melanogaster</i> . <i>Science</i> , 2002 , 298, 149-59	33.3	455

12	Sequential amino acid exchange across b(0,+)-like system in chicken brush border jejunum. <i>Journal of Membrane Biology</i> , 2001 , 180, 213-20	2.3	37
11	Functional analysis of novel mutations in y(+)LAT-1 amino acid transporter gene causing lysinuric protein intolerance (LPI). <i>Human Molecular Genetics</i> , 2000 , 9, 431-8	5.6	56
10	Identification of a membrane protein, LAT-2, that Co-expresses with 4F2 heavy chain, an L-type amino acid transport activity with broad specificity for small and large zwitterionic amino acids. <i>Journal of Biological Chemistry</i> , 1999 , 274, 19738-44	5.4	301
9	Non-type I cystinuria caused by mutations in SLC7A9, encoding a subunit (bo,+AT) of rBAT. <i>Nature Genetics</i> , 1999 , 23, 52-7	36.3	232
8	Identification of SLC7A7, encoding y+LAT-1, as the lysinuric protein intolerance gene. <i>Nature Genetics</i> , 1999 , 21, 293-6	36.3	246
7	Identification and characterization of a membrane protein (y+L amino acid transporter-1) that associates with 4F2hc to encode the amino acid transport activity y+L. A candidate gene for lysinuric protein intolerance. <i>Journal of Biological Chemistry</i> , 1998 , 273, 32437-45	5.4	265
6	The molecular basis of cystinuria: the role of the rBAT gene. <i>Amino Acids</i> , 1996 , 11, 225-46	3.5	13
5	Large-Scale Uniform Analysis of Cancer Whole Genomes in Multiple Computing Environments		14
4	The impact of non-additive genetic associations on age-related complex diseases		3
3	Comparative analysis of neutrophil and monocyte epigenomes		2
2	Human pancreatic islet 3D chromatin architecture provides insights into the genetics of type 2 diabetes		7
1	GCAT Panel, a comprehensive structural variant haplotype map of the Iberian population from high-coverage whole-genome sequencing		1